CASE DISCUSSIONS
MSS (CASES 1-3)
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CASE 1

A 5-year-old boy is brought to clinic for recurrent fractures after minimal trauma. His parents report he fractured his femur at age 2 after falling from standing height. On exam, he has blue sclera, mild hearing difficulty, and joint hypermobility. Radiographs show generalized osteopenia. Genetic testing reveals mutation affecting type I collagen synthesis

Discussion points:

- Q1. What is the most likely diagnosis?
- Q2. What is the underlying molecular defect?
- Q3. Why do patients develop blue sclera?
- Q4. What are the major systemic manifestations?
- Q5. What is the histologic abnormality?
- Q6. What is the major complication affecting prognosis?

Q1. What is the most likely diagnosis?

ANSWER:

Osteogenesis imperfecta

Q2. What is the underlying molecular defect?

ANSWER:

Mutation affecting type I collagen synthesis (COL1A1 or COL1A2 genes) → structurally weak bone matrix.

Q3. Why do patients develop blue sclera?

ANSWER:

Thin sclera → visualization of underlying choroidal veins

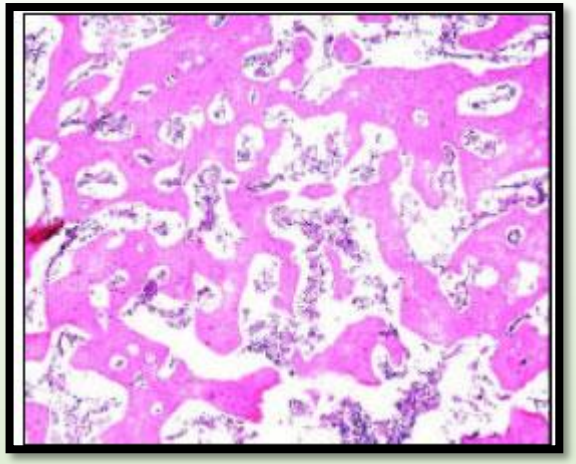
Q4. What are the major systemic manifestations?

ANSWER:

- Fragile bones → recurrent fractures
- Blue sclera
- Hearing loss (abnormal ossicles)
- Dentinogenesis imperfecta
- Ligament laxity

Q5. What is the histological features of OI?

ANSWER:



Reduced and abnormal osteoid with defective collagen framework

Q6. What is the major complication affecting prognosis?

ANSWER:

Repeated fractures and skeletal deformities (severe forms → respiratory failure)

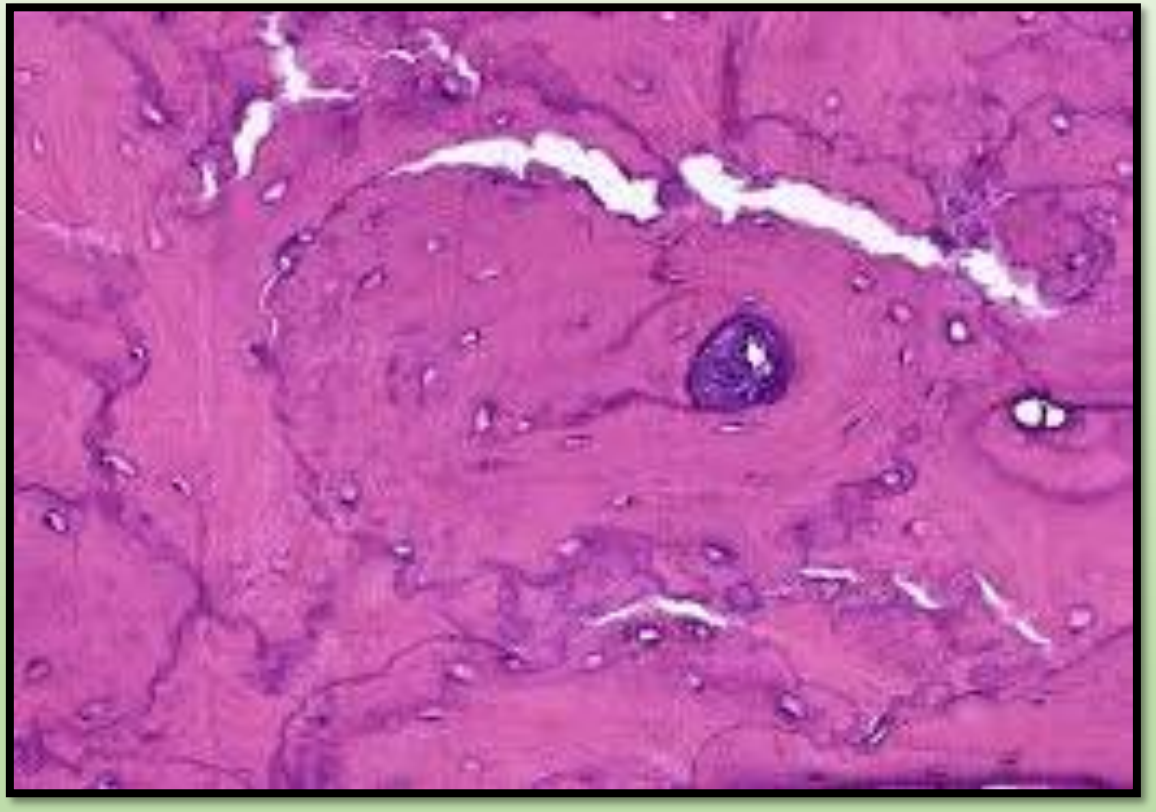
CASE 2

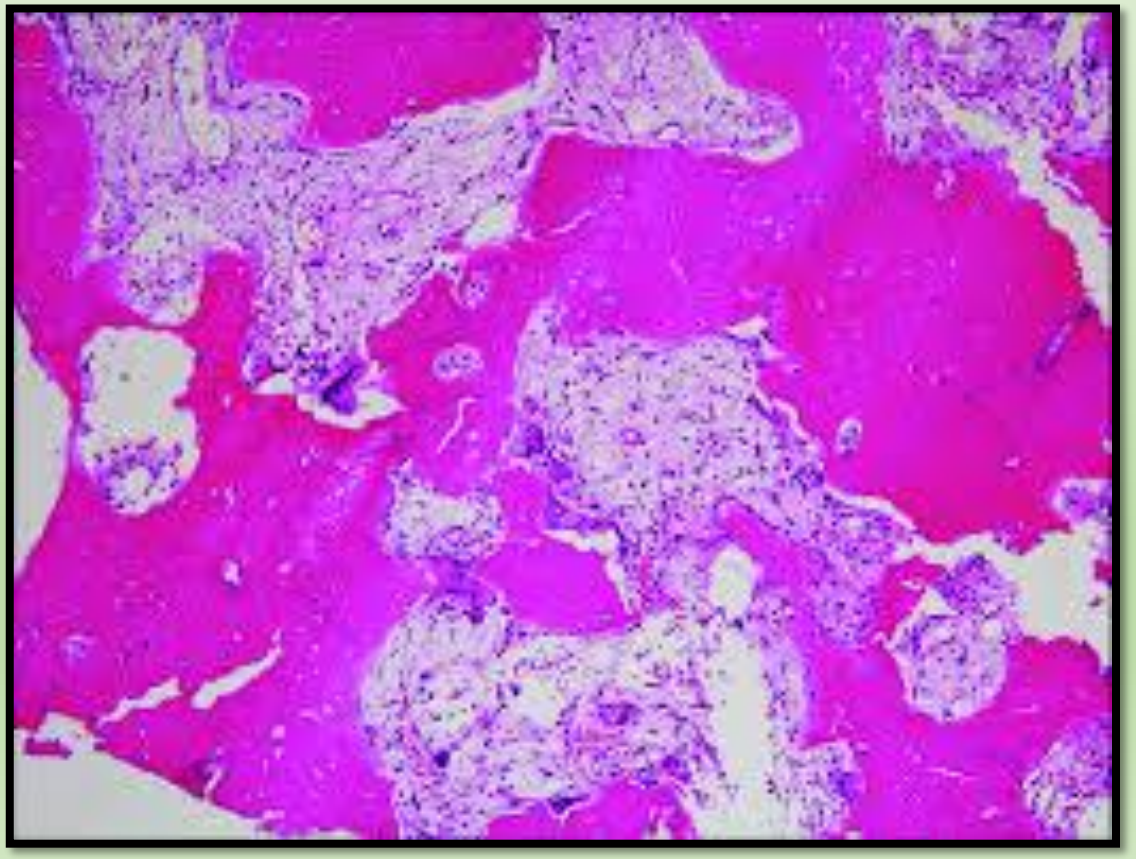
A 68-year-old man presents with increasing hat size and dull skull pain. Labs show markedly elevated alkaline phosphatase with normal calcium and phosphate. X-ray of skull shows thickened bone with patchy sclerosis. Bone biopsy shows mosaic pattern of lamellar bone.

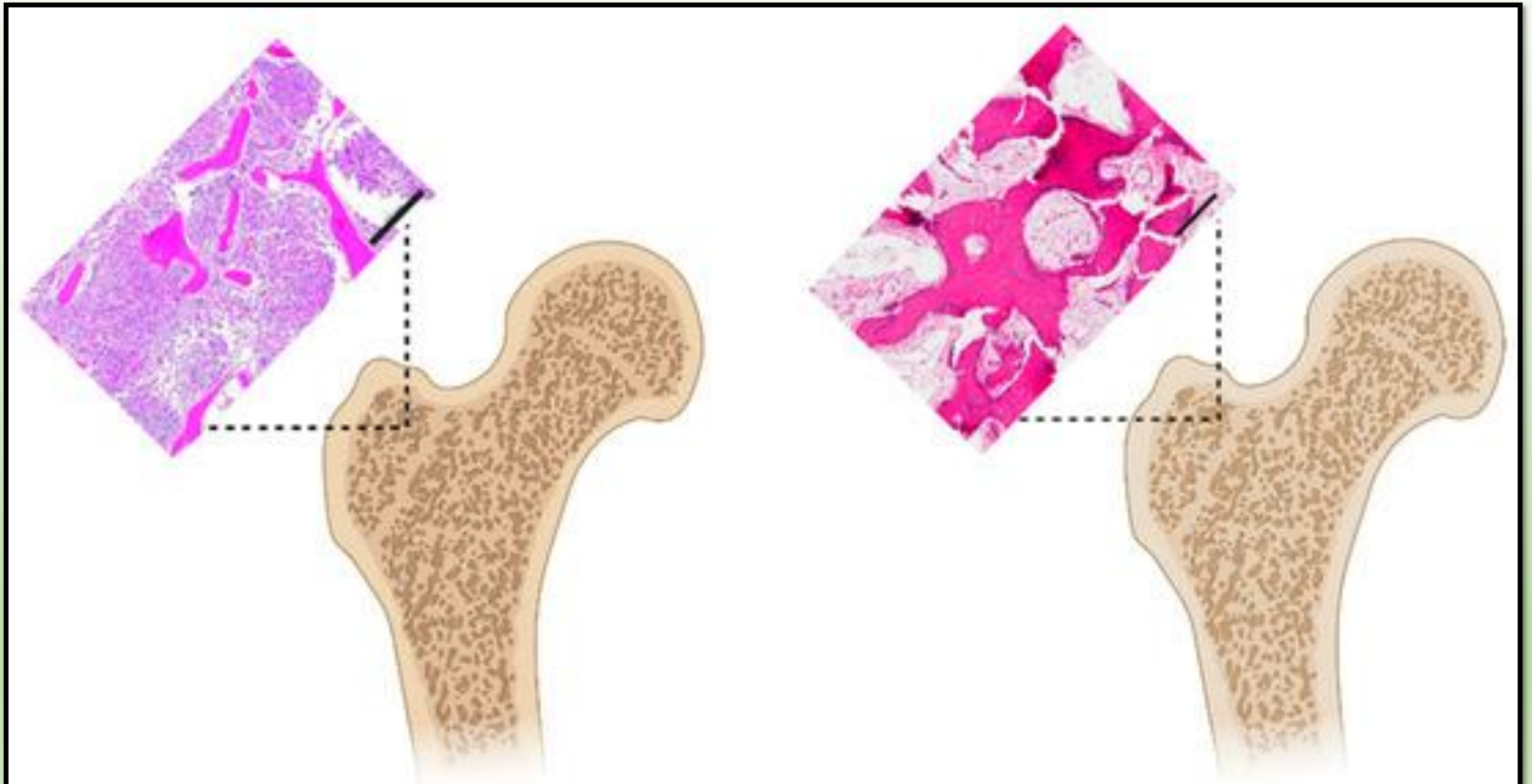
SKULL

PELVIS

HISTOLOGY: MOSAIC PATTERN






Normal Bone
Paget's Disease

DISCUSSION POINTS:

Q1. What is the most likely diagnosis?

Q2. What are the three classic phases of Paget disease?

Q3. Why is alkaline phosphatase elevated?

Q4. What causes the mosaic bone pattern?

Q5. What are important clinical complications?

Q6. Why is calcium usually normal?

Q1. What is the most likely diagnosis?

ANSWER:

Paget disease of bone

Q2. What are the three classic phases of Paget disease?

ANSWERS:

- **Osteolytic phase → ↑ osteoclast activity**
- **Mixed phase → osteoclast + osteoblast activity**
- **Sclerotic phase → disorganized osteoblast bone formation**


LYTIC
SCLEROTIC
MIXED

Q3. Why is alkaline phosphatase elevated?

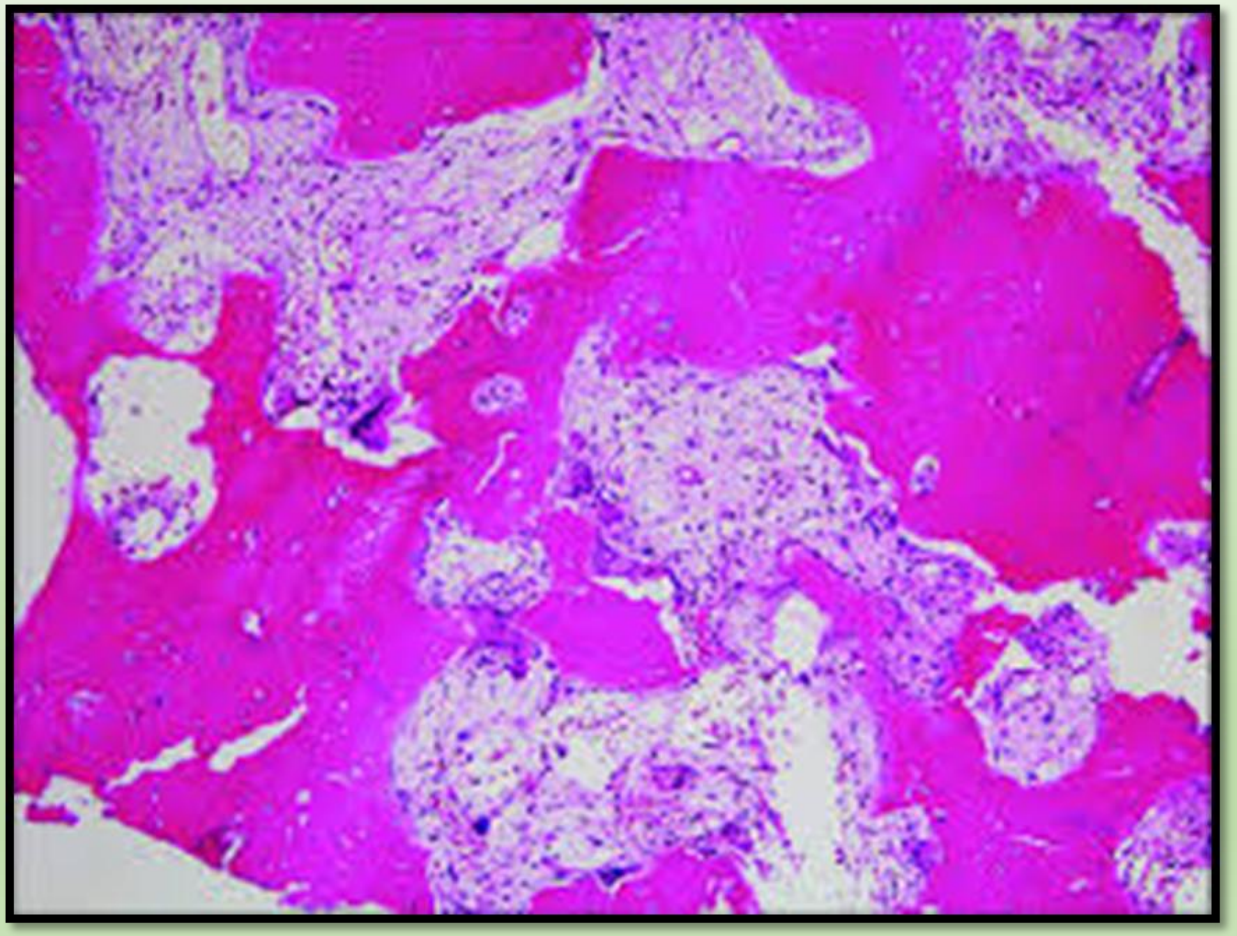
ANSWER:

It reflects increased osteoblastic bone formation activity

Q4. What causes the mosaic bone pattern?

ANSWER:

Disorganized lamellar bone deposition with irregular cement lines.



Q5. What are important clinical complications?

ANSWER:

- Bone deformity
- Hearing loss (skull involvement)
- Pathologic fractures
- Rare: osteosarcoma transformation (long term, rare)

Q6. Why is calcium usually normal?

ANSWER:

Bone turnover is increased but balanced systemically unless immobilized.

Case 3

A 67-year-old postmenopausal woman presents with back pain and progressive loss of height. She has history of vertebral compression fracture after minor trauma. DEXA scan shows T-score –2.9.

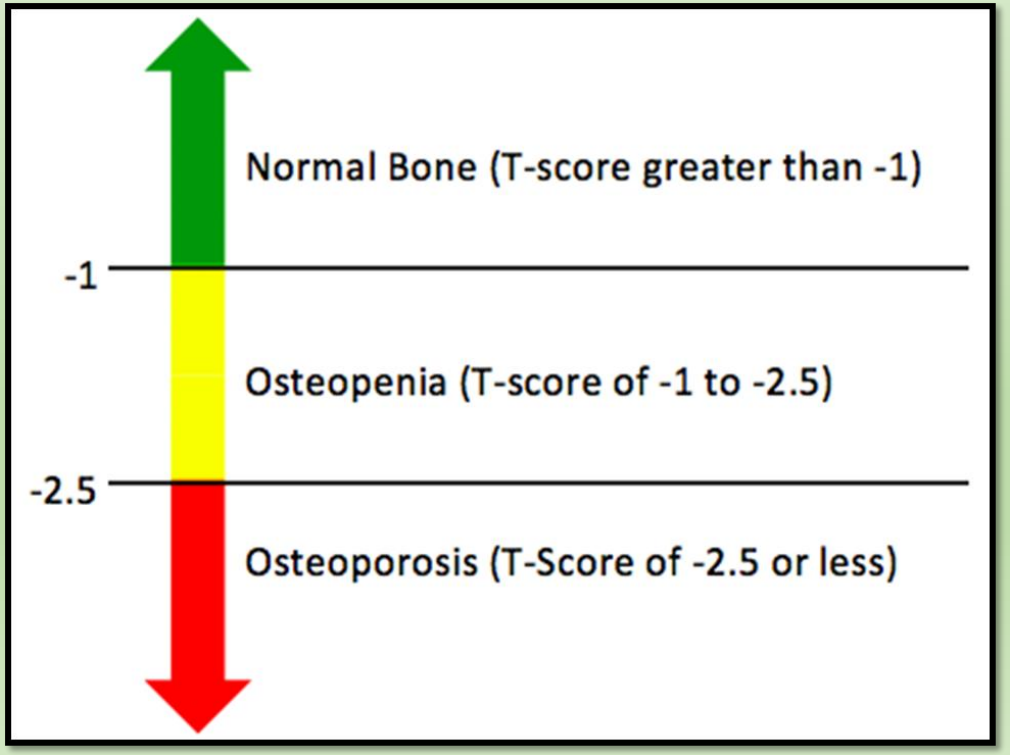
DISCUSSIONS POINTS:

- **Q1. What is the most likely diagnosis?**
- **Q2. What is the WHO diagnostic definition?**
- **Q3. What is the fundamental pathophysiology?**
- **Q4. What are the most common fracture sites?**
- **Q5. What are the major risk factors?**
- **Q6. What is the microscopic appearance?**

Q1. What is the most likely diagnosis?

ANSWER:

OSTEOPOROSIS



Q2. What is the WHO diagnostic definition?

ANSWER:

DEXA scan results:

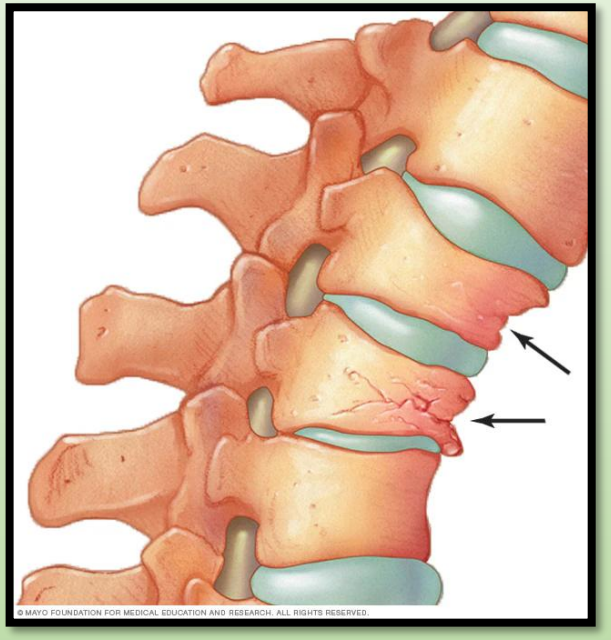
T-score ≤ −2.5

Q3. What is the fundamental pathophysiology?

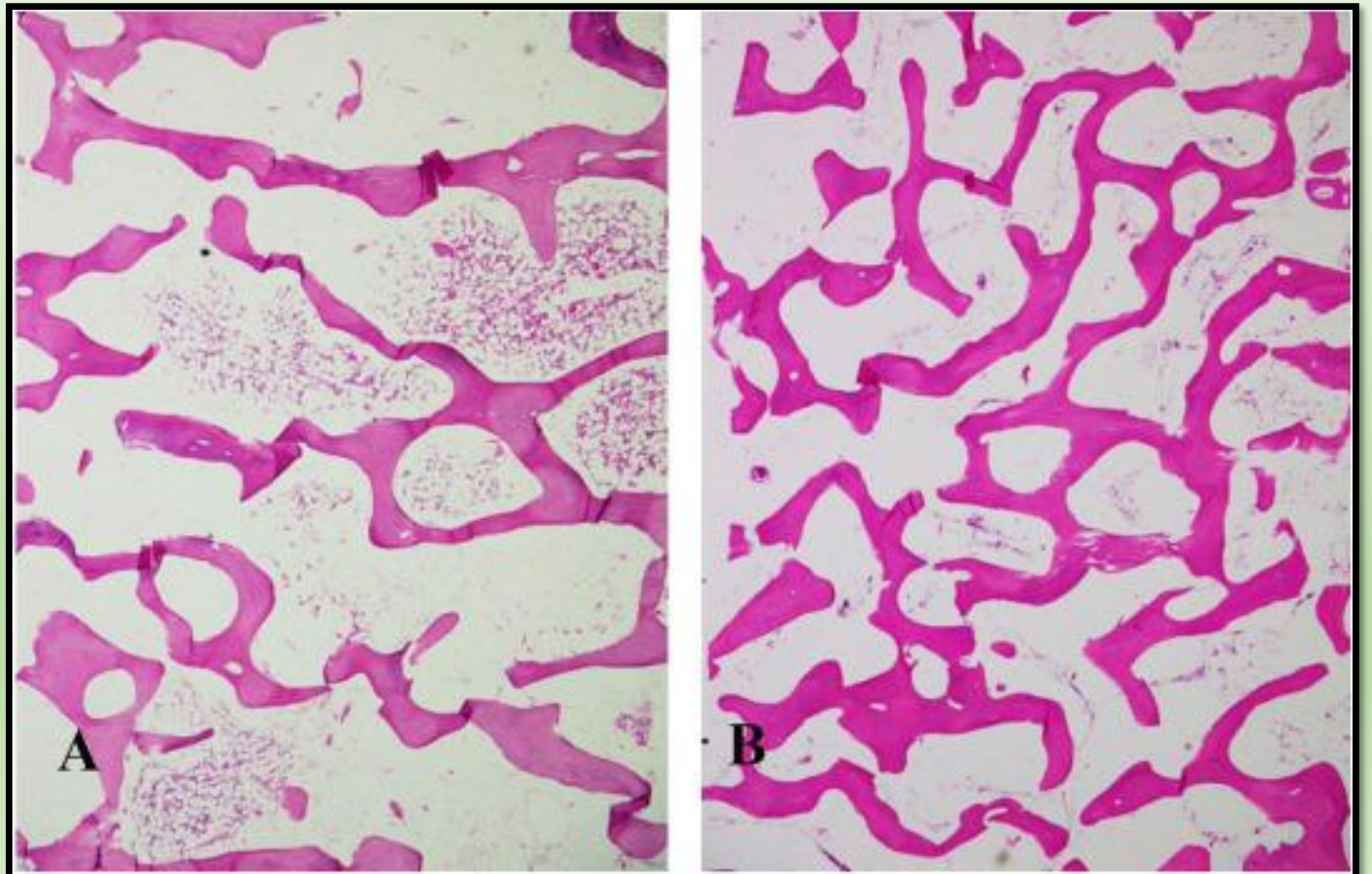
ANSWER:

Bone resorption > bone formation → loss of bone mass and microarchitecture.

Osteoporotic bone shows thinning of trabeculae and cortex, predisposing to fragility fractures.



OSTEOPOROSIS NORMAL



Q4. What are the most common fracture sites?

ANSWER:

- **Vertebral bodies**
- **Femoral neck**
- **Distal radius**

Q5. What are the major risk factors?

ANSWER:

- Postmenopausal estrogen deficiency
- Aging
- Smoking
- Steroid use
- Low BMI
- Vitamin D deficiency
- Sedentary lifestyle

Q6. What is the microscopic appearance?

ANSWER:

Normal mineralization but decreased bone quantity (thin trabeculae, cortical thinning).

OSTEOPOROSIS NORMAL